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Prevalence and Pattern of Congenital Musculoskeletal Disorders among New Born Babies in Port Harcourt, Nigeria.

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ABSTRACT

Studies have shown that musculoskeletal system disorders are one of the most common congenital anomalies. The physical and psychological burden on the child, family, and the society are enormous especially in low-income countries like Nigeria. The aim of this study was to determine the prevalence and pattern of congenital musculoskeletal disorders among new-borns in Port Harcourt. The study analysed all referrals and deliveries during the study period for assessment of any obvious musculoskeletal abnormalities. Data was analyzed using Statgraphics centurion CVI version 16.1.11 and Statistics Package for Social Science. $p<0.05$ was considered statistically significant. Results reveal that 128 new-born had congenital musculoskeletal disorders, with a prevalence of 3.28/1000 births. 88 (68.75%) of all the observed abnormalities involved the lower limbs, with a prevalence of 2.25/1000 births. The most common disorder was congenital talipes equinovarus. In conclusion, Congenital musculoskeletal anomalies have a prevalence of 3.28 per 1000 births. Lower limbs are more affected than other regions of the body, and congenital talipes equinovarus deformity is the most common abnormality seen. Males were more affected than females.

Key Words: Congenital, musculoskeletal, disorders, prevalence, pattern.

INTRODUCTION

Congenital musculoskeletal disorders are defects of the musculoskeletal system recognisable at birth, or later as the baby grows [1,2,3]. They usually occur during intrauterine life, and result from abnormality in the growth and development of soft-tissue structures and bones [4,5]. These disorders include congenital talipes equinovarus (lower limbs), polydactyly (upper and lower limbs), Cleft lip and palate (head), Spina bifida (spine), and Pectus excavatum (thorax) [6,1]. Majority are obvious and usually easy to diagnose while some, such as absent sternum, may be difficult to diagnose [7] except with the aid of radiographs.

About 66% of major congenital anomalies have no recognized aetiology [8]. When the aetiology is known, it is usually multifactorial, involving the interaction of various risk factors such as genetic, environmental, infectious agents, socio-economic/demographic factors, and maternal nutritional status [9]. In-utero exposure to teratogens, such as thalidomide, is a known risk factor of congenital limb defects [10].

A joint World Health Organisation and March of Dimes meeting reports that 7.9 million infants are born yearly having one serious birth defect or the other. And out of this number, 94% (7.4 million) can be seen in middle- and low-income countries [11]. This discrepancy is due to high level of poverty, poor maternal health, increased number of aged mothers, a greater frequency of marriages between close relatives etc. in those countries [12]. The report also showed that about 3.3 million under-five deaths are due to birth defects, with majority of the deaths occurring in middle- and low-income countries [13]. Birth defects, of which congenital musculoskeletal disorders are a major contributor [9,14], are also a significant cause of stillbirths and neonatal mortality [12].

Prevalence studies have shown that the musculoskeletal system disorders are the most common congenital anomalies consisting about 29.2% of all congenital anomalies [15]. Some other studies observed a prevalence of 20/1000 live births [16], and 30/1000 live births [17]. Eventually, when these musculoskeletal disorders are not corrected, the children often develop lifelong deficits. These come with enormous functional, psychological, educational, and financial burden on both the affected children and their families [18]. Many of these children end up not attending school [19].

The physical and psychological burden of Congenital Musculoskeletal Disorders on the child, family, and the society, are enormous. This burden is highest in low-

income countries like Nigeria, where congenital musculoskeletal disorders are more prevalent[13]. Despite the impact of these disorders on the child and society, the risk factors, pattern, and prevalence have not been sufficiently studied, and most of the data are not country specific. In response to these concerns, we conducted this study to evaluate the prevalence and pattern of congenital musculoskeletal disorders among new-borns in Port Harcourt

MATERIALS AND METHODS

This retrospective study was conducted in two tertiary health care settings in Port Harcourt, Nigeria. The clinical records of the new born who presented with musculoskeletal anomalies within the study period (1st January 2019– 31st of December 2020) were reviewed for various congenital musculoskeletal disorders, and risk factors associated with severity of these disorders.

During this period, a total of 9, 635 cases were reviewed, out of which 128 new-borns were identified with musculoskeletal anomalies. A medical record review checklist was used to obtain information on socio-demographic data [20] such as sex of the babies, age of mother, number of pregnancies and deliveries, educational level of mother, family history of musculoskeletal anomalies, exposure to chemicals and irradiation, alcohol abuse, use of recreational drugs etc. Information about the frequency of occurrence, side of the body affected and bilaterality, sex distribution of the anomalies, description of the anomalies and diagnosis were also obtained.

The data was cleaned and analysed using STATGRAPHICS centurion CVI version 16.1.11 (StatPoint Tech., Inc.) and Statistics Package for Social Science (SPSS) – (IBM® Amos V21.0.0, USA). The descriptive statistics was performed using STATGRAPHICS and the categorical variables distribution were presented as frequencies (%), while continuous variable was presented as mean (±S. D). The prevalence was calculated and pattern determined. Chi Square was used to test the association between maternal age and musculoskeletal disorders, and the differences in the occurrence of congenital musculoskeletal disorders in males and females. $P < 0.05$ was taken as significant.

RESULTS

Out of a total of 9,635 of cases seen, 128 newborns were found to have congenital musculoskeletal disorders. The congenital musculoskeletal disorders comprise 26 different types of disorders involving different regions of the body. The various congenital musculoskeletal disorders and their distribution are illustrated in Figure 2.

88 (68.75%) of all the observed abnormalities involved the lower limbs. The most common disorder was Congenital talipes equinovarus (CTEV) seen in 49 (38.28%) of the newborn (Table 1). Other observed abnormalities of the lower limbs include polydactyly seen in 9 (7.03%) of the new born, genu valgum, genu varum, syndactyly, metatarsus adduction, pes planus, and talipes calcaneovalgus, in their order of frequency. CTEV was more common in the males (33) than in the females (16), and more common bilaterally than unilaterally. When it occurred unilaterally, it was more common on the right than on the left (table 1).

Head abnormalities were the second most common abnormalities, seen in 23 (17.97%) of the newborn. Congenital hydrocephalus was the commonest head abnormality, seen in 5 (3.91%) of all the newborn, followed by cleft palate and meningomyelocele (Table 1). 75 (58.59%) out of the 128 newborns were males, while 53 (41.41%) were females (Table 1), but a test of difference in the occurrence of musculoskeletal disorders in Males and Females was not significant (Table 4).

Table1: Pattern and sex distribution of Congenital musculoskeletal disorders. n=128

S/N	TYPE OF DISORDER	Total Number	LIMB		SIDE			SEX		%
			Upper	Lower	RIGHT	LEFT	Both	M	F	
1	Congenital talipes equinovarus	49		49	8	2	39	33	16	38.28
2	Polydactyly	9	4	5	4	2	3	4	5	7.03
3	Genu valgum	7		7	1	--	6	2	5	5.46
4	Genu varum	5		5	--	--	5	4	1	3.90
5	Congenital hydrocephalus	5						3	2	3.90
6	Syndactyly	5	2	3	3	2		2	3	3.90
7	Spina bifida	5						3	2	3.90
8	Cleft palate	4						3	1	3.13
9	Meningomyelocele	4						3	1	3.13
10	Metatarsus Adduction	4		4	2	1	1	2	2	3.13
11	Scoliosis	4						1	3	3.13
12	Pes planus	4		4	--	--	4	2	2	3.13
13	Microcephaly	3						1	2	2.24
14	Cleft lip	3						1	2	2.34
15	Talipes Calcaneovalgus	3		3	2	1	--	2	1	2.34
16	Congenital hip dysplasia	2		2			2	1	1	1.56
17	Encephalocele	2						1	1	1.56
18	Rocker bottom foot	2		2			2	1	1	1.56
1 9	Absent distal Interphalangeal Joint	1	1		1			1		0.78
20	Absent sternum	1						1		0.78
21	Arthrogryposis	1					1	1		0.78
22	High arched palate	1						1		0.78
23	External tibial torsion	1		1		1			1	0.78
24	Genu recurvatum	1		1			1		1	0.78
25	Hypoplastic femur	1		1	1			1		0.78
26	Flattened occiput	1						1		0.78
Tota l		128	7	88	22	9	64	7 5	53	

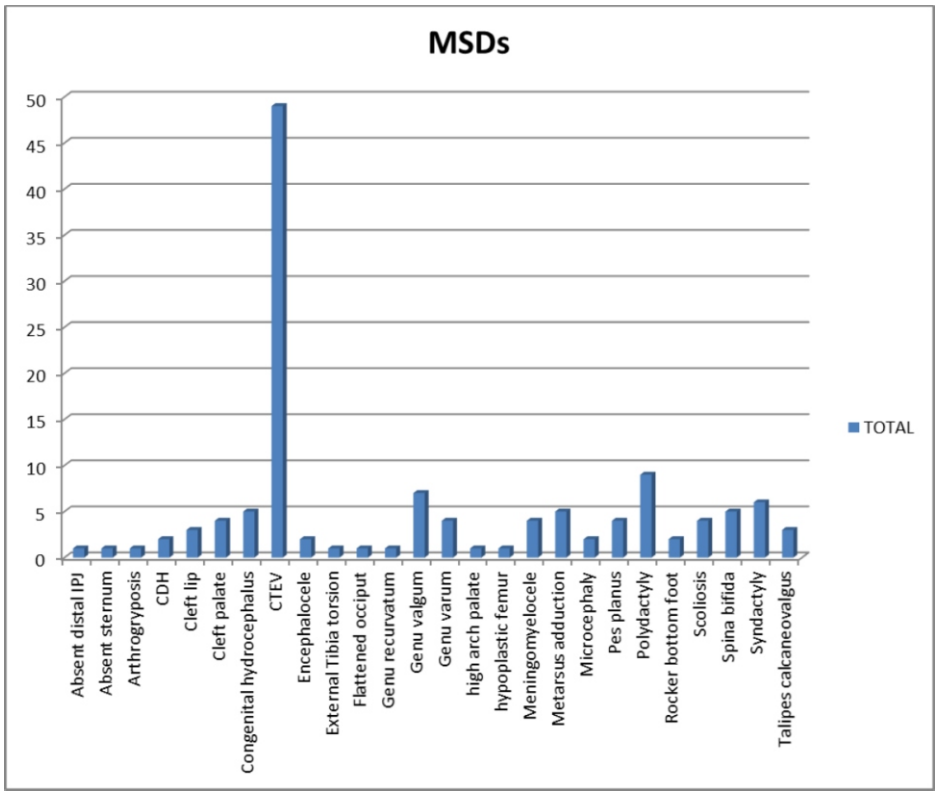

Figure 1: Distribution of musculoskeletal disorders

Table 2: Regional distribution of congenital musculoskeletal disorders.

S/NO	REGION OF THE BODY	FREQUENCY	%
1	Upper Limb	7	5.47
2	Lower Limb	88	68.75
3	Spine	10	7.81
4	Head	23	17.97

Table 3: Prevalence of musculoskeletal disorders

	TOTAL BIRTHS (UPTH+RSUTH)	CONGENITAL MSDs	PROPOR-TION	%	PREVALENCE/ 1000 BIRTHS
2019	24,461	80	0.0033	0.33	3.27/1000 Births
2020	14,569	44	0.0030	0.30	3.29/1000 Births
OVERALL	39,030	128	0.0033	0.33	3.28/1000 Births

Table 4: Test of differences in the occurrence of MSDs in Males and Females

S/N	TYPES OF MSD DISORDER	FEMALES	MALES	Chi square value	P value	Inference
1	Absent distal Interphalangeal joint	0	1	7.36	0.99	Not significant
2	Absent sternum	0	1			
3	Arthrogryposis	0	1			
4	Congenital dysplasia of hip	1	1			
5	Cleft lip	2	1			
6	Cleft palate	1	3			
7	Congenital hydrocephalus	2	3			
8	Congenital talipes equinovarus	16	33			
9	Encephalocele	1	1			
10	External Tibia torsion	1	0			
11	Flattened occiput	0	1			
12	Genu recurvatum	1	0			
13	Genu valgum	5	2			
14	Genu varum	1	4			
15	high arch palate	0	1			
16	hypoplastic femur	0	1			
17	Meningomyelocele	1	3			
18	Metatarsus adduction	2	2			
19	Microcephaly	2	1			
20	Pes planus	2	2			
21	Polydactyly	5	4			
22	Rocker bottom foot	1	1			
23	Scoliosis	3	1			
24	Spina bifida	2	3			
25	Syndactyly	3	2			
26	Talipes calcaneovalgus	1	2			
	TOTAL	53	75			

*$p < 0.05$ is considered significant.

Maternal age was quite variable and ranges between 24-42 years with a mean of 32.7years, and Mean±S.D of 32.7±3.708. A test of association between musculoskeletal disorders (MSDs) and maternal age was not significant (Table5).

Table 5: Test of Association between MSDs and maternal Age

S/N	TYPE OF MSD DISORDER	20-30 years	31-40 years	>40 years	Chi Square Value	P-value	Inference
1	Absent distal IPJ	0	1	0	33.26	0.97	Not Significant
2	Absent sternum	0	1	0			
3	Arthrogryposis	0	1	0			
4	CDH	2	2	0			
5	Cleft lip	0	1	0			
6	Cleft palate	0	4	0			
7	Congenital hydrocephalus	2	4	0			
8	CTEV	18	30	1			
9	Encephalocele	1	1	0			
10	External Tibia torsion	0	1	0			
11	Flattened occiput	0	1	0			
12	Genu recurvatum	0	1	0			
13	Genu valgum	5	2	0			
14	Genu varum	3	1	0			
15	high arch palate	1	0	0			
16	hypoplastic femur	0	1	0			
17	Meningomyelocele	2	2	0			
18	Metarsus adduction	1	4	0			
19	Microcephaly	0	2	0			
20	Pes planus	1	2	0			
21	Polydactyly	4	5	0			
22	Rocker bottom foot	0	2	0			
23	Scoliosis	1	3	0			
24	Spina bifida	3	2	0			
25	Syndactyly	3	2	1			
26	Talipes calcaneovalgus	0	3	0			
TOTAL		47	79	2			

*p< 0.05 is considered significant. IPJ= Interphalangeal joint. CDH=congenital dysplasia of the hip
CTEV= Congenital Talipes Equinovarus

DISCUSSION

This study investigated the prevalence and pattern of musculoskeletal disorders among new born in Port Harcourt. The prevalence is 3.28/1000 births in Port Harcourt City of Nigeria. Saib *et al* [21] reported a prevalence of 9.4/1000 births in South Africa, while Kumari & Singh [22] reported a prevalence of 7.01/1000 births in the Indian population. These results are comparatively higher than ours, and studies by Adane *et al* [23] who reported a prevalence of 3.9/1000 births in the Sub-Saharan region and Ghorpade *et al* [6] who reported a prevalence of 2.03/1000 births in India. For the 2years study in Port Harcourt, the yearly prevalence was 3.27/1000 births in 2019 and 3.29/1000 births in 2020. The slight decrease in 2019 prevalence when compared to 2020 could be attributed the aversion people had towards the hospital environment as a result of the COVID-19 pandemic which was at its peak in 2019.

The difference in the prevalence observed in this study when compared to other studies can be explained by the robustness of our study. This current study included patients born in the hospitals where the study was conducted, and those that were referred from peripheral

centres. Ghorpade *et al* [6] assessed only patients born in the hospital, and hence, reported a lower prevalence when compared to that of our study. Also, our study did not include miscarriages and still births, so this may be a reason for a lower prevalence when compared to studies that yielded a high prevalence. Another reason for the difference in the prevalence could be as a result of the design of the study. The current study was a hospital-based study, and one would expect a higher prevalence of congenital musculoskeletal abnormalities in a setting where sick new born, with possible congenital anomalies, are treated as compared to a community-based study.

Again, our study recorded a higher prevalence when compared to that recorded in high-income countries. Figueroa *et al* [24] conducted a study in Guatemala, central America, and reported a lower prevalence of 1.08/1000 births. This higher prevalence reported in our study when compared to that reported in high-income countries could be due to the differences in the practice of prenatal diagnosis. In these high-income countries, the practice of prenatal diagnosis is a routine, so some of the congenital anomalies are diagnosed prenatally and eventually aborted. This could account for the low prevalence in these high-income countries. This lack of practice of prenatal diagnosis in low-income countries can be attributed to poverty, ignorance, and lack of requisite expertise/experience for prenatal diagnosis. Furthermore, the risk factors of congenital musculoskeletal disorders such as folic acid deficiency, use of unprescribed medications, exposure to waste sites, exposure to infectious agents, etc. are more common in low-income countries. This also, could account for the higher prevalence reported in these countries.

Eighty-eight (88) of all the observed abnormalities involved the lower limb (68.75%), with CTEV (Club foot) as the commonest disorder, seen in 49 (38.28%) of the newborn. There was also a predominance of bilateral involvement when compared to unilateral involement. CTEV was followed by polydactyly, seen in 9 (7.03%) of the newborn. This also, is comparable with studies in different parts of Nigeria. Orimolade *et al* [26] reported a similar pattern in their study in which they observed that lower limb abnormalities were the most common, with CTEV deformity seen in 55% of patients followed by polydactyly (13%). They also reported a predominance of bilateral involvement in CTEV. Edomwonyi *et al* [14] reported a somewhat similar pattern with CTEV as the commonest (61.3%) lower limb abnormality. However, in their report, CTEV was followed by syndactyly (26,7%) instead of polydactyly as reported in our study. Other studies from outside the country showed a pattern similar to our study. Ghorpade *et al* [6] reported that 94 of all the 217 identified abnormalities involved the foot, with CTEV as the most common, which was seen in 60 neonates. This was followed by polydactyly.

Report of studies done in the western countries differ from that of our study, and other Nigerian studies, as they showed that disorders of the upper limb were more common than that of lower limbs. A study carried out a study in Italy reported that 64% of patients had upper limb anomalies, while 21% had lower limb anomalies [27].

The fact that these studies were conducted in different geographical locations and at different times could explain the reason for the difference in the pattern of occurrence of congenital musculoskeletal disorders. This could also be attributed partly to different sampling techniques, inclusion and exclusion criteria, genetic constitution, and environmental factors which play an important role in the aetiopathogenesis of congenital abnormalities [28]. However, there is need for further investigation of these differences.

In this study, 75 (58.59%) were males while 53 (41.41%) were females, with a male: female of 1.4:1. International studies, such as that done by Kumari & Singh [23] and Aigoro *et al* [29] reported a similar predominance of congenital musculoskeletal disorders in males when compared to females, and a similar male to female ratio. However, other studies reported a predominance of musculoskeletal disorders, especially neural tube defects (NTDs), in females [30]. The exact reason for this difference is unknown, however some studies suggested the role of p53 activation on the X-chromosome [31]. The study found that p53 binds to the X-chromosome inactivation center (XIC) and activates the XIC genes. If this binding does not take place, there is a probable failure of X-chromosome inactivation. The study concluded that X chromosome inactivation failure may be the reason for female bias in neural tube closure defects. It has been observed that p53 plays a vital role in conserving stability by preventing genome mutation [32].

The current study reveals that children with congenital musculoskeletal disorders are more among mothers aged 31–40 years, although we found no correlation between congenital musculoskeletal anomalies with increase in maternal age. This agrees with findings by Sarkar *et al* [33]. However, some other studies reported a significant correlation between congenital musculoskeletal anomalies with increase in maternal age [34,6].

CONCLUSION

Congenital musculoskeletal disorders have a prevalence of 3.28/1000 births. The lower limb is the most affected region of the body, with CTEV as the most common disorder seen. This prevalence and pattern vary from one region to the other, and from time to time. To reduce the morbidity and long-term consequences associated with these anomalies, corrective measures have to be instituted early.

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